Biochimica et Biophysica Acta 1792 (2009) 823

Contents lists available at ScienceDirect

Biochimica et Biophysica Acta

journal homepage: www.elsevier.com/locate/bbadis

Preface

From glycosylation to glycosylation diseases

This volume on Congenital Disorders of Glycosylation (CDG) appears 10 years after the magnificent BBA volume on the Molecular Basis of Glycoconjugate Diseases edited by Harry Schachter [1]. We are very happy and proud that this charming "pope" of glycobiology has in turn agreed to contribute to the present volume. In these 10 years, CDG (formerly called Carbohydrate-deficient Glycoprotein Disorders) have come of age. In 1999, only 6 CDG were known: in a mutase (CDG-Ia), an isomerase (CDG-Ib), a glucosyltransferase (CDG-Ic), a mannosyltransferase (CDG-Id), an *N*-acetylglucosaminyltransferase (CDG-IIa), and a galactosyltransferase (*B4GALT7* defect). This number has increased to more than 40 by now including also defects in glycosidases, chaperons, oligosaccharyltransferase, transporters, a kinase and a flippase (see synoptic table in [2]). Besides the known defects in protein *N*-glycosylation and protein *O*-glycosylation, new subfamilies have emerged in particular the combined protein *N*- and *O*-glycosylation defects, dolichol metabolism defects, glycosylphosphatidylinositol anchor defects and glycolipid synthesis defects. Moreover, genetic diseases due to hyperglycosylation have been recognized [3].

Besides a large repertoire of neurological and other symptoms, an increasing number of known specific syndromes have been identified as glycosylation disorders. Examples of these are the hereditary multiple exostoses, the congenital muscular dystrophies, Peters plus syndrome, leukocyte adhesion deficiency type II, autosomal recessive hereditary inclusion body myopathy, the allelic disorders hyperphosphatemic familial tumoral calcinosis and cortical hyperostosis with hyperphosphatemia, spondylocostal dysostosis type 3 and cutis laxa type II.

Over those 10 years, we have been witnessing a growing and fruitful international collaboration on research in CDG. At the same time, the awareness has been increasing, to the extent that more patients are being identified worldwide, year after year. These do not only include the typical and known cases but also lead to a growing list of unsolved cases. The latter then become the subject of further research promoting our insights in glycosylation and its diseases. The medical and rare disease community has seldom seen such close interactions between clinical and basic research. Two European projects, Euroglycan and its successor, Euroglycanet, have significantly contributed to the success. And now that the funding by the European Commission is drawing to its end, a larger international collaboration is likely to emerge because, also in the USA, a diagnostic and research network on CDG has been formed.

Finally, we would like to use the publication of this compilation of papers on CDG to honour a colleague and friend who has spent much of his research career on the study of glycosylation. It is a great pleasure to dedicate this special issue to Eric Berger on occasion of his retirement as professor of physiology at the University of Zürich.

We cordially thank Paul Fraser and Ben Oostra, the executive editors of *Molecular Basis of Disease* for the invitation to organize this volume, and the authors, referees and the staff of Elsevier for their contributions.

References

[1] H. Schachter, Molecular basis of glycoconjugate disease, Biochim. Biophys. Acta (1455) (1999) 61–62.
[2] J. Jaeken, T. Hennet, H.H. Freeze, G. Matthijs, On the nomenclature of congenital disorders of glycosylation, J. Inherit. Metab. Dis. 31 (2008) 669–672.
[3] G. Vogt, B. Vogt, N. Chuzhanova, K. Julenius, D.N. Cooper, J.L. Casanova, Gain-of-glycosylation mutations, Curr. Opin. Genet. Dev. 17 (2007) 245–251.

Jaak Jaeken
Center for Metabolic Diseases, University Hospital Gasthuisberg, Herestraat, 49, BE-3000 Leuven, Belgium

Gert Matthijs
Center for Human Genetics, University Hospital Gasthuisberg, Herestraat, 49, BE-3000 Leuven, Belgium

Dr. Jaak Jaeken is an emeritus professor of pediatrics at the University of Leuven, Belgium. He received his M.D. degree from the University of Leuven in 1967. From 1967 to 1973 he trained in pediatrics at the same university. Subsequently he completed a fellowship in metabolic diseases at the University of Zürich in the laboratory of Dr. Richard Gitzelmann. In 1975 he returned to Leuven as a clinical investigator with main topics such as disorders in the metabolism of ammonia, amino acids, neurotransmitters and purines, and the congenital disorders of glycosylation which he first described in 1980. He received his Ph.D. in 1985. In 1999 he received the degree of Doctor Honoris Causa from the University of Zürich, and in 2000 from the University of Havana.

Gert Matthijs (°1963), Ph.D., is the head of the Laboratory for Molecular Diagnostics at the Center for Human Genetics in Leuven, and professor at the University of Leuven, Belgium. He is a molecular geneticist, involved in the diagnostics of inherited diseases since 1994. His major research interest is in Congenital Disorders of Glycosylation (CDG), a group of rare inborn errors of metabolism. He is the coordinator of Euroglycanet, a European project focusing on the identification of novel defects and the generation of mouse models for CDG. For this work on CDG, he received the "Körber European Science Award" in 2004, together with Prof. von Figura, Prof. Aebi, Prof. Hennet, Prof. Jaeken and Prof. Lehle. He is the deputy-coordinator of Eurogentest, a network of excellence (NoE) for development, harmonization, validation and standardization of genetic testing in Europe, funded by the European Commission. He was also actively involved in the European opposition against the BRCA patents. At the national level, he has been a driving force for a revision of the reimbursement system for genetic tests.

0925-4439/$ – see front matter
doi:10.1016/j.bbadis.2009.08.003